Harlequin ichthyosis (HI, OMIM 242500) is a rare and most severe form of autosomal recessive congenital ichthyoses characterized by excessive hyperkeratosis. The skin anomalies affect the shape of eyes, ears, nose, mouth, and distal extremities. Since HI is very fatal, there is a lack of data that documents successful treatment of HI patients with syndactyly. We successfully managed syndactyly in a HI patient by using a conventional method without any complication and we noticed an improvement functionally and aesthetically.

**Key Words:** Lamellar ichthyosis, Syndactyly, Hand deformities, Skin transplantation

Harlequin ichthyosis (HI) is a rare se- vere form of autosomal recessive congenital ichthyoses and is usually fatal. It is characterized by excessive hyperkeratosis resulting in thick scales separated by deep, erythematous and diamond-shaped fissures in the skin. HI is caused by recessive mutations in the gene encoding the adenosine triphosphate-binding cassette transporter protein ABCA12 (NCBI Entrez Gene 26154). This gene is localized to the lamellar granules of upper epidermal keratinocytes. ABCA12 plays a key role in keratinocyte differentiation by expressing markers that participate in late epidermal differentiation. The skin anomalies affect the shape of eyes, ears, nose, mouth and distal extremities. Syndactyly is one of features shown in HI patient.

Syndactyly is one of the most common congenital anomalies of the upper limb and several surgical techniques with skin graft has been implemented with great results. However, surgical outcomes for syndactyly patients with HI are not well known due to the early mortality rate of the patient population. Also, syndactyly in HI patient is associated with skin contracture and consequent angular deformity. Herein, we introduce a case that treats HI patients with syndactyly surgically.

**CASE REPORT**

A 6-year-old female patient visited an outpatient clinic with syndactyly in bilateral hand and foot. On physical
examination, all the web spaces in hand and foot were involved (Fig. 1). Skin contracture and ulnar deviation of in proximal, middle, distal interphalangeal (IP) joints and metacarpophalangeal (MCP) joint were noted. Also, flexion contracture, active and passive limitation of extensors were noted in proximal, middle, distal IP joints, and MCP joints (Fig. 2). These features were related to characteristics of HI patient. The patient was unable to grasp objects with her hand despite prolonged physical therapy. To improve her hand function, surgical intervention was determined. The decision was made to perform surgery on the second web space of right hand. A dorsal rectangular flap and a volar triangular flap were designed (Fig. 3). After the division of second and third finger, a dorsal rectangular flap and a volar triangular flap flaps were interposed to recreate the second web. After interposition of flaps, the raw surface was covered with full thickness skin sized 4×2 cm harvested from the left inguinal crease (Fig. 4). Postoperatively, compressive dressing with gauze and an elastic bandage was maintained for 2 weeks. Interposed flaps healed well and full thickness skin grafts were taken perfectly. After 6 months, reconstructed web space was maintained without recurrence (Fig. 5). Six months follow up X-ray image showed significant improvement of ulnar drift. The ulnar deviation angle at MCP joint of second finger corrected from 60 to 25 degree (Fig. 6). In addition, functional improvements were evident by the patient being able to manipulate the joystick of her electric wheelchair and grasp paper money. However, patient underwent minor wound dehiscence in left inguinal donor site, which managed revision and moisturization (Fig. 7).

**DISCUSSION**

There is a lack of clinical outcome of syndactyly correction in HI patients. However, longer life expectancy are now achieved with the improvement of care for HI patients⁴. Since keratinocyte-fibroblast interaction is one of the key steps in wound healing⁸, dysregulation of epidermal differentiation in HI patients might have negative
impact on wound healing. So we concerned of wound healing after surgery. However, functional and aesthetic aspects were improved after surgery. This might suggest that angular contracture of HI patients might be secondary to skin contracture instead of joint problems. So we suggest that adequate release of syndactyly and contracture is important in HI patient. So in HI patients, standard surgical options for syndactyly could be adjustable. In addition, we would like to recommend carefully design the flaps to minimize the raw surface for skin graft due to insufficient skin laxity for full thickness skin donor site. Additionally, one of important postoperative management is moisturizing of recipient and donor site. Moisturizing in HI patients might promote wound healing and contribute to skin contracture release and soften skin scale. Overland and Johnstone\textsuperscript{9} previously reported hand manifestations of HI patient treated with full thickness skin graft and had no complications. We expect that full-thickness skin graft can be safely performed on HI patient. In conclusion, we successfully managed syndactyly in an HI patient with full thickness skin grafts without any complication and saw improvement functionally and aesthetically.

CONFLICT OF INTEREST

The authors have nothing to disclose.
Fig. 5. Postoperative 6-month photograph. Right second web space is well-healed without complication. (A) Top view. (B) Bottom view. (C) Tip view.

Fig. 6. Postoperative X-ray of right hand. Angle of ulnar deviation of metacarpophalangeal joint of second finger is 25 degree.

Fig. 7. Postoperative wound dehiscence in donor site. At postoperative 2 weeks, left inguinal area underwent minor wound dehiscence which required surgical revision.
REFERENCES


할리퀸 어린선에서 합지증의 수술적 치료

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할리퀸 어린선은 과각화증으로 대표되는 상염색체 열성 선천성 어린선 중 가장 희귀하고 심각한 형태이다. 이러한 피부의 변형은 눈, 귀, 코, 입, 그리고 사지에도 영향을 미친다. 할리퀸 어린선은 대개 치명적으로, 할리퀸 어린선 환자의 합지증 치료에 대한 보고가 거의 없다. 본원에서는 할리퀸 어린선 환아의 합지증을 기존의 합지증 수술법을 이용하여 큰 합병증 없이 치료하였고, 기능적이고 미적인 향상을 관찰하였다.

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